Sgrinio Smotyn Gwaed Newydd-anedig Cymru
Newborn Bloodspot Screening Wales

GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

Newborn bloodspot screening programme

Your baby's screening result

A sickle cell disorder (SCD) is suspected

Your baby's screening result

The result of your baby's 'heel prick' screening blood test suggests they might have a sickle cell disorder. Your baby now needs further tests to check whether he or she has a sickle cell disorder or not.

This leaflet gives you some information about sickle cell disorders and explains what happens next.

What are sickle cell disorders?

Sickle cell disorders are conditions in which the red blood cells can become shaped like a sickle or crescent moon ('sickled cells'). Red blood cells contain haemoglobin, which is responsible for carrying oxygen from the lungs to the tissues in the body. With sickle cell disorders, the red blood cells contain haemoglobin S instead of the usual haemoglobin A. This causes the red blood cells to change shape when the cell has released its oxygen. Sickled red blood cells are not as flexible as normal-shaped blood cells and can become stuck in the small blood vessels. This can cause pain, anaemia, damage to tissue and infection.

During the first three to six months of life, your child may not show signs of having a sickle cell disorder because at birth there is a high level of baby haemoglobin (haemoglobin F). The haemoglobin F prevents the red blood cells from changing shape. Over the first year of life the haemoglobin F levels become lower, but some children continue to make higher levels of haemoglobin F even into adulthood.

Newborn bloodspot screening means that babies with SCD can be identified and receive early treatment before problems are likely to develop.

Treatment

For most babies with a confirmed sickle cell disorder, treatment involves health monitoring, medication, education and support throughout their life to avoid and prevent the complications of sickle cell disorder as much as possible. This allows children to lead as active, healthy and fulfilled a life as possible. Children with a sickle cell disorder will be under the care of a specialist medical team.

The treatment is usually with regular antibiotics to prevent infection, and it is important that your child has their routine vaccinations.

What happens next?

You will be given an appointment to see a specialist medical team who will:

- discuss the screening test result with you
- arrange for your baby to have a blood test to confirm the diagnosis
- support you now and in the future if your baby has a confirmed diagnosis of a sickle cell disorder
- let your family doctor know about your baby's test results
- give you written information about sickle cell disorders to share with your family and medical professionals if your baby needs medical care, and
- answer any questions you may have.

Your questions answered

What happens if my baby becomes unwell before our appointment with the specialist medical team?

If your baby becomes unwell you should get medical advice quickly. It is important that the healthcare professional you contact is aware of this screening result for sickle cell disorders and we suggest that you show them this leaflet.

Why do some children have sickle cell disorders?

Sickle cell disorders are inherited conditions. They are not caused by anything that happened during pregnancy.

A baby has two copies of the haemoglobin gene. In a baby born with a sickle cell disorder, neither copy of the haemoglobin genes is working correctly because both genes have an alteration (also known as a 'gene variant'). This alteration affects the structure of haemoglobin.

What is life like for children with sickle cell disorders?

Sickle cell disorders vary greatly in how severe they are and it is difficult to predict how the disorder will affect an individual. Quality of life can be improved if the condition is identified early and treated with antibiotics. Education and support for the family and access to care by a specialist medical team also help improve quality of life.

Will other members of the family also have a sickle cell disorder?

If a baby is confirmed to have a sickle cell disorder, the risk to the parents of having another child together with sickle cell disorder is 1 in 4 (25%) for every child. Parents or other family members can be referred to specialist genetic services for more information and advice if they want to.

More information and support

- You can find the booklet ‘A parent’s guide to managing sickle cell disease’ on the Sickle Cell Society website: www.sicklecellsociety.org/resource/parentsguide/

- The Sickle Cell Society: www.sicklecellsociety.org

- Newborn Bloodspot Screening Wales: phw.nhs.wales/newborn-bloodspot-screening

Notes

You can write down any notes here from discussions with your health professionals.

Using your information

For us to contact you as part of the programme, we will need to handle some of your and your baby's personal information. If you need more information about this, you can:

- visit the website: phw.nhs.wales/use-of-site/privacy-notice
- email PHW.InformationGovernance@wales.nhs.uk, or
- phone 029 2010 4307.

We also keep personal details to make sure that the standard of our service is as high as possible. This includes checking your baby's records if your baby is found to have a condition after having a screening test which showed a 'not suspected' result.

We only ever publish information as statistics and we never publish personal details. We pass on your personal information to health professionals or organisations that need it, including your GP, health visitor and consultant paediatrician. These professionals must protect the personal information in the same way as we do.

All our paper and computer records are stored and processed securely, and the public do not have access to them.

Information courtesy of Public Health England.

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